

Picture of the month

## Predominant distal muscle involvement in spinal muscular atrophy

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Spinal muscular atrophy (SMA) is an autosomal recessive disorder characterized by degeneration of alpha motor neurons, clinically associated with muscle weakness, hypotonia and muscle atrophy. The weakness is predominant in the proximal muscles, with lower limbs often more involved than upper limbs with a typical distribution [1]. In the most severe cases the involvement is more diffuse affecting both proximal and distal muscles with very limited movements of hands and feet. The predominant proximal involvement has also been observed on muscle Magnetic Resonance Imaging (MRI) [2,3].

We report the case of a 9-year-old boy with genetically confirmed type 2 SMA. The onset was after 9 months. At that time the child was able to sit independently but was unable to bear weight on his legs and had absent tendon reflexes. This prompted to perform genetic analysis that showed homozygous deletion of exon 7 in the SMN1 gene and 3 SMN2 copies. At the last follow up, at the age of

9 years, both clinical examination and muscle MRI showed a predominant involvement of the leg muscles compared to the thigh muscles. On clinical examination, there was generalized weakness, more marked in the distal lower limbs. Muscle MRI T1 sequences were acquired on axial plane covering pelvis, thigh and leg. Muscle MRI showed a diffuse involvement of all leg muscles with minimal sparing of the extensor digitorum. At thigh level the involvement was overall less diffuse with relative sparing of adductor longus, gracilis and semitendinosus (Fig 1a). This pattern is different from that observed in other SMA patients who show the more typical proximal to distal gradient (Fig 1b) or a diffuse involvement (Fig. 1c).

Our results suggest that, in addition to the well known patterns of involvement observed in SMA patients, a predominant involvement of the distal muscles can also be found. Further studies in larger cohorts will help to establish the recurrence of this pattern.

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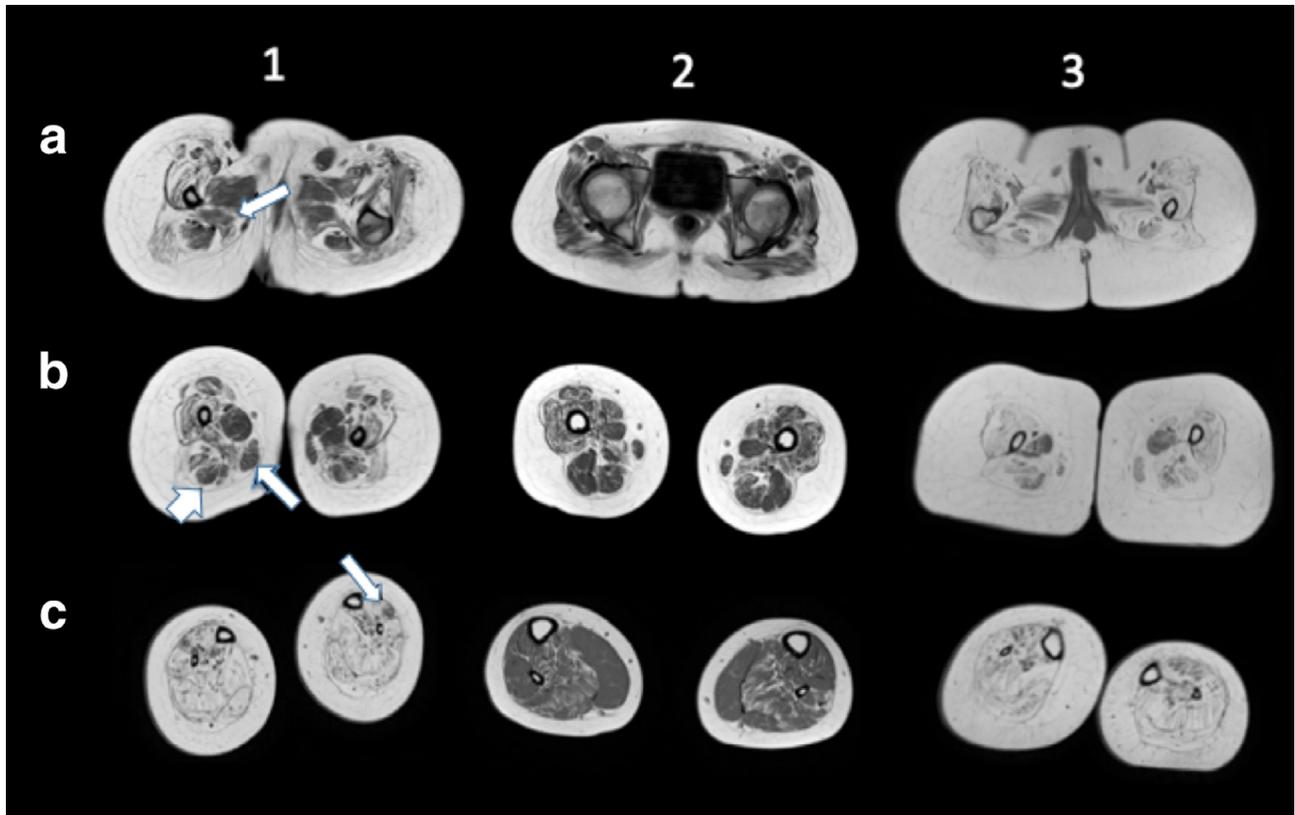


Fig. 1. Predominant distal muscle involvement in SMA. T1 weighted TSE images acquired on axial plane through different levels: row A: pelvis, row B: thigh, row C: leg. The images show that in patient 1 there was more distal than proximal involvement. At pelvis level (1a) relative sparing of adductor muscles (arrow) can be seen; at thigh level (1b) adductor longus, gracilis (arrow) and semitendinosus (arrowhead) are the less involved muscles; at leg level (1 c) diffuse hyperintensity suggestive of fatty replacement can be seen in all muscles except extensor digitorum longus (arrow). The pattern of involvement in patient 1 was different from the typical patterns observed in two other patients with similar genetic diagnosis and similar age of onset. In patient 2, a 7 year old boy, the involvement was more proximal than distal, as classically reported for SMA patients. In patient 3, a 16 year old boy, there was an overall diffuse involvement, associated with a severe generalized weakness.

## References

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